



GEN52 - Management of the vascular abnormalities associated with vascular Ehlers–Danlos syndrome

OBJECTIVE: To retrospectively review the natural history of vEDS and describe the clinical outcomes of patients who have undergone surgical repair for vascular events and those who are undergoing surveillance

ORGANIZATION

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BACKGROUND AND RATIONALE

Vascular EDS is an autosomal dominant disorder resulting from mutations in the COL3A1 gene encoding type III procollagen. Excessive tissue fragility in individuals with vEDS may result in rupture or dissection of a previously non-dilated artery, although arterial aneurysms are also seen in this population. While the natural history of vEDS has been well described, no long term data regarding the effectiveness of endovascular repair or surgical guidelines for that management of patients with vEDS exists.

DESIGN

Method: Retrospectively review the natural history of vEDS and describe the clinical outcomes of patients who have undergone surgical repair for vascular events and those who are undergoing surveillance. The long term objective is to develop recommendations and guidelines regarding appropriate surveillance frequency and surgical approaches for patients with vEDS.

Inclusion criteria:

- Subjects with vEDS

Samples:

- None

Data:

- Demographics
- Imaging
- Medication Use
- Surgical
- Organ system review
- Genetic

CONCLUSIONS

Results:

- *Aortic and mesenteric arterial involvement in vEDS appears to be related to the underlying mutation type, with the HI cases having milder disease and later presentation. Molecular diagnosis is warranted in vEDS cases as it predicts postoperative outcomes and guides surveillance.*

